

A novel NIR spectroscopic qualitative method to monitor the diet compliance in PKU patients.

POSTER n°69 - Theme 3: Share, Care, Rare: Transforming care for rare diseases by 2030

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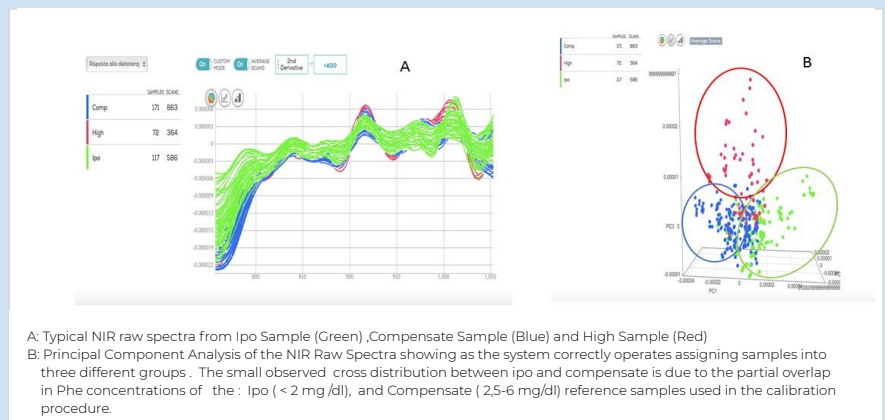
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Introduction

Phenylketonuria (PKU MIM 261600) is a human metabolic disease, caused by mutations in the phenylalanine hydroxylase gene. Children born with PKU, thanks to the newborn screening and early dietary interventions, can have relatively normal lives. The diet allows them to keep Phe level in range. The best indicator of dietary compliance is regular monitoring of Phe concentrations by using a Liquid Chromatography (LC) based procedure. We developed a qualitative chemometric model able to separate PKU patients based on their response to the diet in three different categories: **Low** (<2 mg/dl), **Compensate** (2,5-6 mg/dl) and **High** (6,5- 20 mg/dl) with NIR spectroscopy.

Methods and results

A qualitative chemometric model was elaborated using 3000 NIR spectra, from 562 DBS PKU samples from the three different groups, based on their Phe levels assessed by LC. Using a sensor calibrated in the Near infra-Red spectrum, 20 scans per sample in duplicate were acquired. Principal component analysis (PCA) was conducted. The chemometric model was validated by blind external procedure. Our model is able to correctly assign unknown sample to the Low, Compensate, or High group with a linearity of 0,91 and a $\sigma=0,078$, showing a sensitivity ratio between the number of the correctly classified samples (CCS) and the number of the known samples belonging to each class (KS): $Sn=CCS \cdot 100KS$ close to 100%.

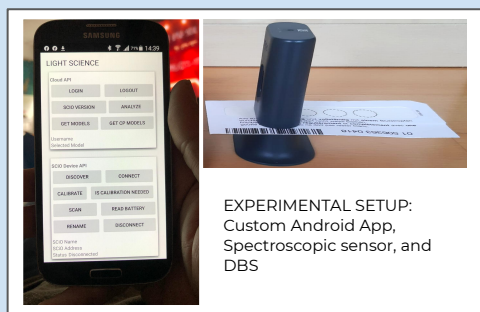


Target: Risposta alla dietoterapia
 Performance: F1 = 0.9511

Classified Known Class	Comp	Ipo	High
Ipo	0%	100%	0%
High	0%	0%	100%
Comp	100%	0%	0%

Model Info
 Target: Risposta alla dietoterapia
 Performance: F1 = 0.9511
 Algorithm: RF
 Outlier Detection: ON
 Cross Validation By: Sample
 Number of Folds: 5
 Min Scans Per Sample: 5 (Average)

Predictive Chemiometric Full model for Ipo, Compensate and High samples
 Performance evaluation of the predictive chemiometric model for CS vs I and PS obtained by matrix distribution analysis. The estimated performance value is 0.951, with only 4% of false compensate and no bias for the others two categories



Conclusions

We demonstrated that it is possible to use NIR spectroscopy to quickly monitor the diet compliance in PKU patients.